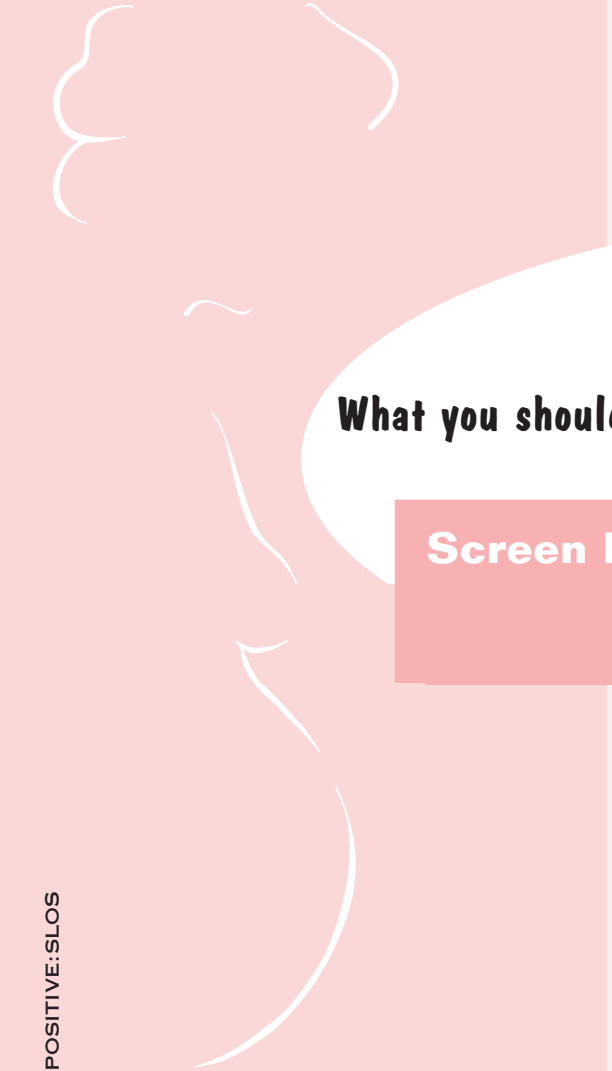


What you should know when your Expanded AFP blood test result is

Screen Positive: Indicating an increased risk for SLOS/Other Birth Defects

POSITIVE:SLOS

The California Expanded AFP Screening Program

THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

THE CALIFORNIA DEPARTMENT OF HEALTH SERVICES, GENETIC DISEASE BRANCH
850 MARINA BAY PARKWAY, F175, RICHMOND, CA 94804-6403
(866) 718-7915

As part of your prenatal care you had the Expanded AFP blood test. This screening test helps detect some birth defects such as Down syndrome, neural tube and abdominal wall defects, trisomy 18, SLOS (Smith- Lemli-Opitz syndrome), and other abnormalities.

The blood test measured three substances in your blood: AFP (alpha-fetoprotein), HCG (human chorionic gonadotropin) and UE (unconjugated estriol). All of these substances are normally found in a woman's blood when she is pregnant.

Your test result was "screen positive" based on a very low amount of UE in your blood. This indicates an increased risk (chance) there is a pregnancy complication or birth defect or that the fetus may have SLOS.

What could have caused your "screen positive" result?

Many times, the reason for this result is **NOT** a birth defect or pregnancy complication. Sometimes the low level of UE occurs for no known reason.

To help determine ***why your result was "screen positive"***, you are being offered diagnostic follow-up services at a State-approved Prenatal Diagnosis Center. There is no additional charge for approved services.

What can diagnostic tests show?

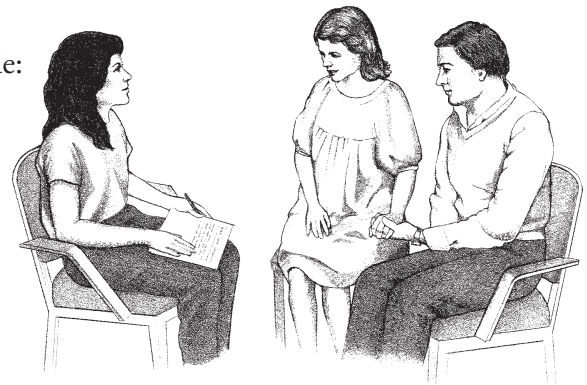
Ultrasound can show some pregnancy complications or birth defects. An amniocentesis can diagnose chromosome abnormalities and fetal cholesterol problems such as SLOS.

Many women with "screen positive" results will have normal follow-up tests and healthy babies.

What are the follow-up services offered at a State-approved Prenatal Diagnosis Center?

These follow-up services include:

- genetic counseling
- ultrasound
- amniocentesis



Counseling will give you information to help you make decisions.

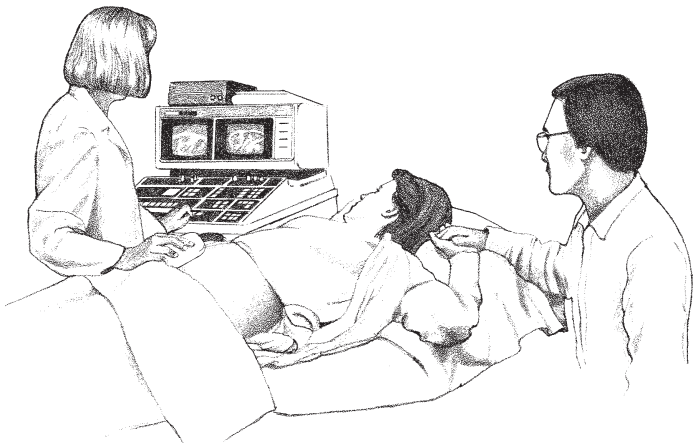
Genetic counseling:

Prenatal Diagnosis services begin with genetic counseling. A genetic counselor or doctor will discuss your blood test result and what it could mean. Your family's health history will be reviewed. Counseling will give you information to help you make decisions about having the follow-up tests. Be sure to talk with the counselor or doctor about any questions or concerns you may have.

Ultrasound:

This test is also called a sonogram. Sound waves are used to make a picture of the fetus. This picture is seen on a special TV screen. Ultrasound does not hurt the baby. Ultrasound can show how the pregnancy is progressing.

It shows the age of the fetus and how many fetuses there are. It also may show whether there is a visible abnormality in the fetus or the uterus.



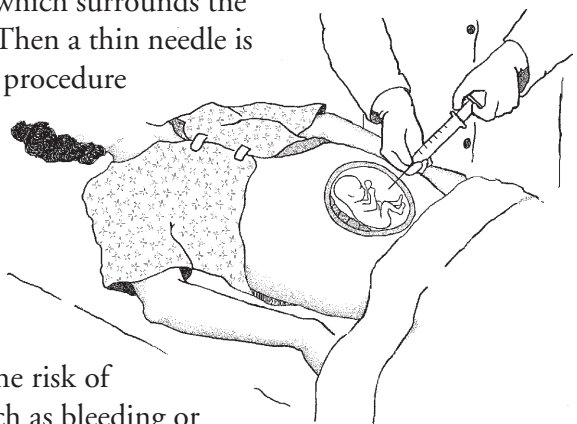
Amniocentesis is offered . . . .

- ***if ultrasound does not explain the "screen positive" blood test result,***

or

- ***if ultrasound finds a problem which needs clarification.***

Amniocentesis:



This test involves removing a small amount of the amniotic fluid which surrounds the fetus. First, an ultrasound picture locates the fetus and the fluid. Then a thin needle is used to remove a small amount of the fluid from the uterus. This procedure sometimes causes brief discomfort. The amniotic fluid contains fetal cells. The chromosomes in these cells are counted and examined. A special test for cholesterol metabolism in the fetus is also done to test for SLOS.

The results of amniocentesis are usually ready in two weeks.

Amniocentesis is considered a safe procedure when performed by medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage following amniocentesis is less than 1%. Problems such as bleeding or infection are rare.

Women can accept genetic counseling and ultrasound, and still decline amniocentesis.

Sometimes not all of the follow-up tests are needed. Genetic counselors will explain which tests will be useful.

Many amniocentesis results are normal.
Sometimes, however, chomosome abnormalities or birth defects such as SLOS are diagnosed.

Smith-Lemli-Opitz Syndrome (SLOS)

Babies with SLOS cannot make enough cholesterol to help their brains and bodies develop normally. Infants born with this birth defect may have severe mental retardation and have numerous health problems. SLOS only occurs in about appoximately 1 out of 100,000 births in the United States.

If the fetus does have SLOS or other birth defects, will the follow-up tests detect it?

Amniocentesis detects almost 100% of SLOS and chromosome abnormalities.

What if the follow-up tests show a problem with the pregnancy or a birth defect?

If a birth defect or pregnancy complications are found, a doctor or genetic counselor will give the woman or couple information about the problems and how the fetus may be affected. Available treatments and options for continuing or ending the pregnancy will be discussed.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

Please remember:

Many women who have had a "screen positive" test result will have normal healthy babies. However, in those pregnancies with a serious birth defect or pregnancy complication, early detection allows parents to explore their options and make early decisions.

The goal of the Genetic Disease Branch is to provide high-quality, low-cost prenatal screening services to all pregnant women in California. If you have questions, comments or suggestions about services received through the California Expanded AFP Screening Program, please let us know.

Write to:

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